

## Opis choroby \*

### Definicja

An exceedingly rare, autosomal recessive immune disease characterized by thumb aplasia, short stature with skeletal abnormalities, and combined immunodeficiency described in three sibships from two possibly related families. The skeletal abnormalities included unfused olecranon and the immunodeficiency manifested with severe chickenpox and chronic candidiasis. No new cases have been reported since 1978.

### Dane

### Klasyfikacja

Zespół wad wrodzonych

#### Kod ORPHA

2951

#### Kod OMIM

274190

#### Kod ICD10

D82.8

#### Kod ICD11

4A01.1Y

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### \*Źródło

orphanet